San Francisco
September 9, 2004

Foundational Issues in Biomedical Ontologies

Workshop

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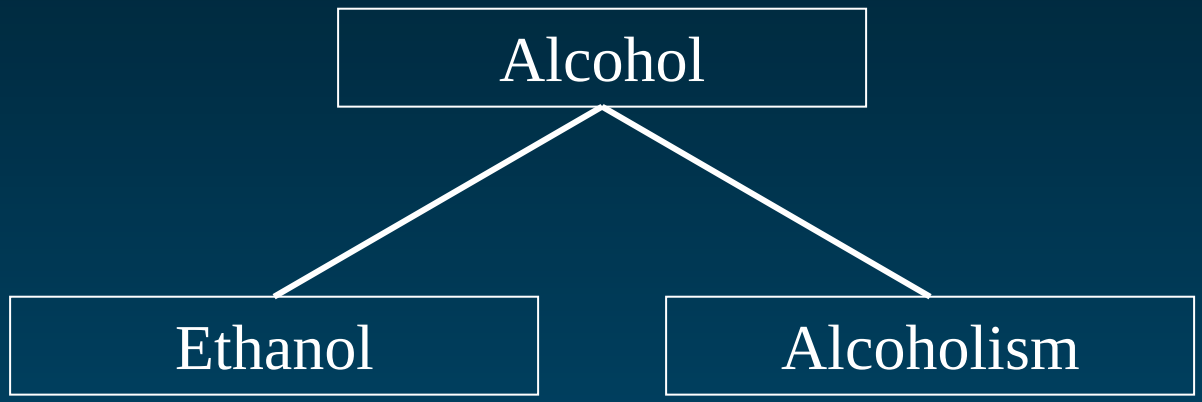
4. The proper treatment of is-a, part-of and other foundational relations and of top-level categories such as function, role, process, structure, activity

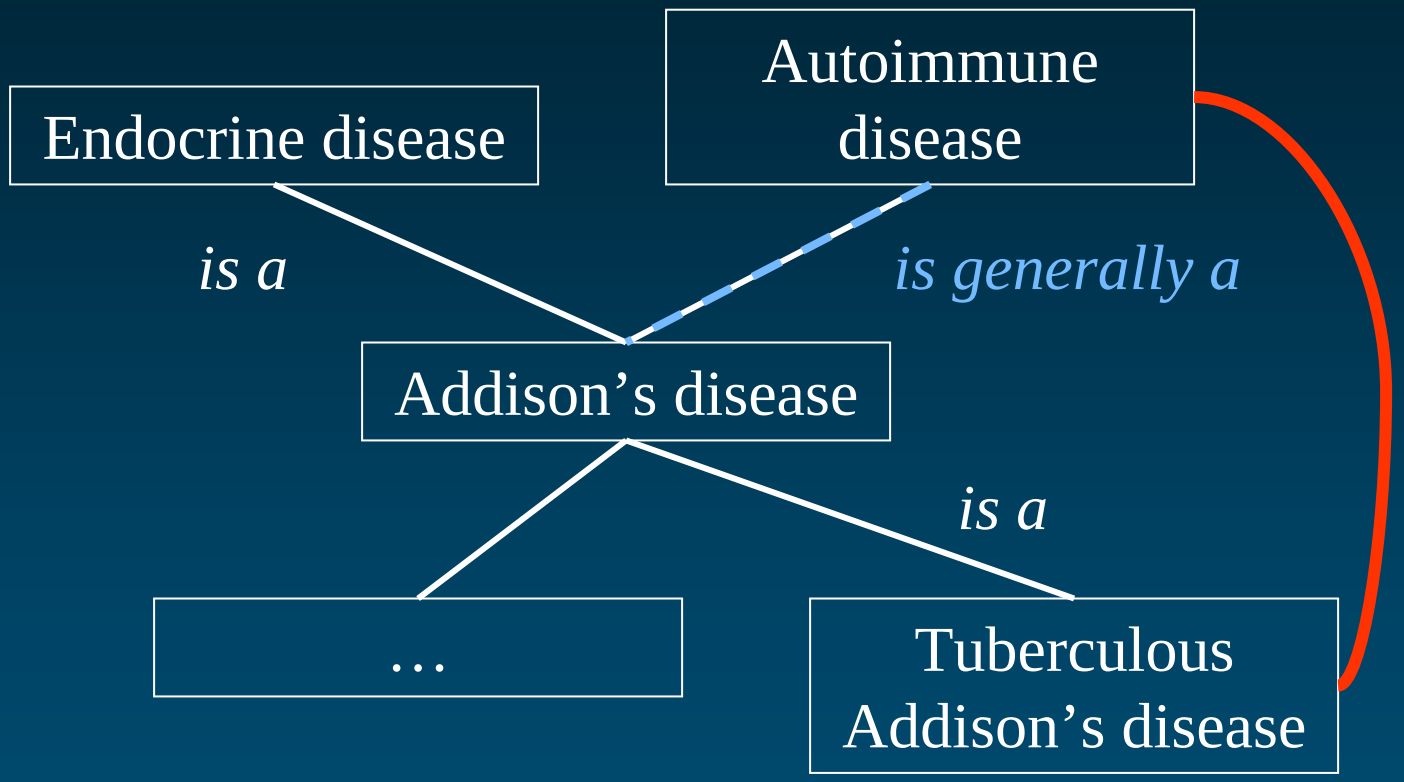
Biomedical vocabularies vs. Biomedical ontologies

- Biomedical vocabularies
  - Objective: information retrieval
  - Thesaurus model
  - Hierarchies: *parent/child*, *broader/narrower than*
  - Not every relation is labeled
  - Not every relationship is precisely defined
- Biomedical ontologies
  - Objective: inference, reasoning
  - Ontology model
  - Hierarchies: hierarchical relationships
  - Every relation is labeled
  - Every relationship is precisely defined

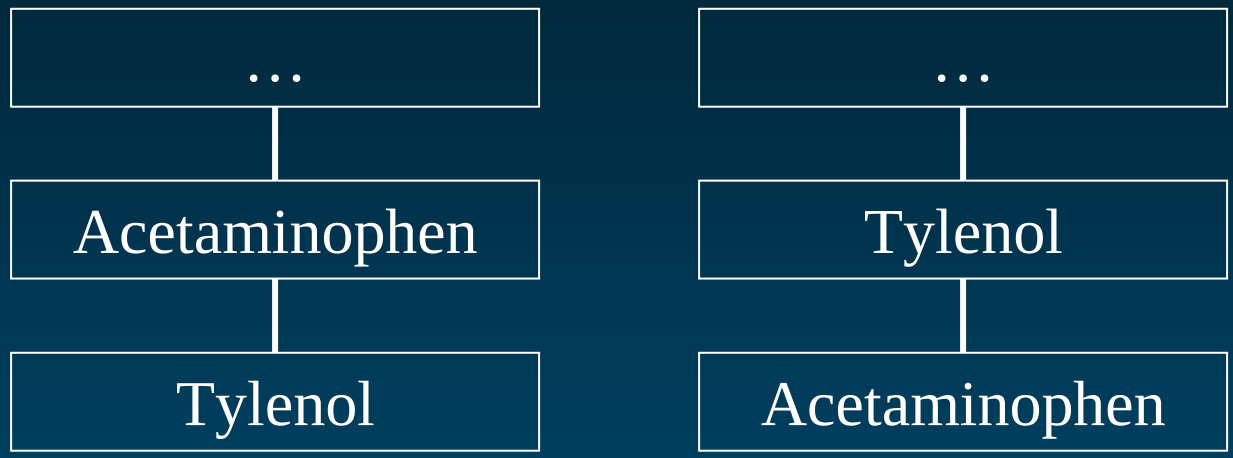
Thesaurus relationships

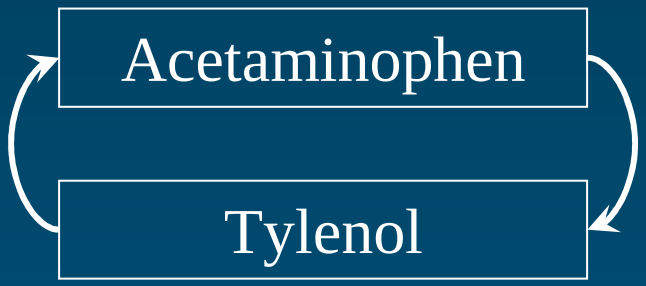


Imprecise definition of relationships



Hierarchies vs. Hierarchical relations





5. The proper treatment of classes and instances (including prototypical, variant and borderline instances)

Classes vs. Instances What

- Classes
  - Represent
    - Kinds of things
    - [Abstractions]
  - Have
    - Instances (mandatorily)
    - Subclasses (possibly)
  - Have “vertical” relations
    - Superclass of subclass
    - Has instance instance
  - Have “horizontal” relations to other classes

- Instances
  - Represent
    - The things themselves
  - Have “vertical” relations
    - Is instance of Class
    - NB: an instance cannot have instances
  - Have “horizontal” relations to other instances

Classes vs. Instances Where

- Biomedical terminologies and ontologies contain mostly classes
  - Exceptions: few geographical areas, laws, persons
- Column names in databases, field names in forms

- Clinical databases and micro-array experiments contain instances
- Values in databases can be either classes or instances

Classes vs. Instances

- A fine-grained class does not make it an instance
- A leaf cannot be either a class or an instance
  - *Subclass of*: Relationship between classes
  - *Is instance of*: Relationship between an instance and a class
- In some systems, it does not matter whether leaf classes are implemented as subclasses or as instances

6. The proper treatment of epistemological concepts such as finding, measurement, and test results in medical ontologies

Ontology vs. Epistemology

- Ontology
  - What exists (in reality)
    - Regardless of how we know it
    - Regardless of modality
  - Breast cancer
- Epistemology
  - What exists, through
    - How we came into being it existed
    - Modality
  - Probable breast cancer
  - Breast cancer detected during a routine mammography

Epistemology in Biomedicine

- Epistemology is everywhere
  - Beside the diagnosis, doctors may record how they came to the diagnosis
    - To explain of the diagnostic strategy
    - For accountability reasons
- Incomplete, most probable diagnosis

Epistemology in biomedical terminology

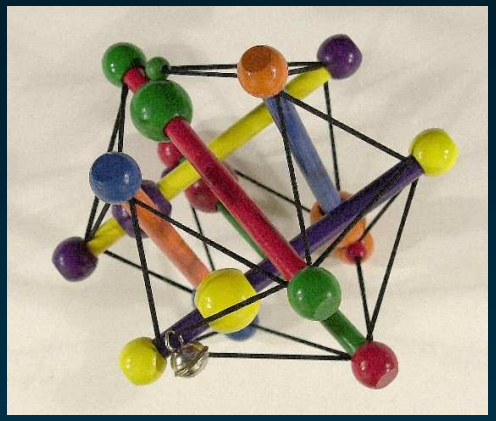
- Epistemological features built in terms
  - *Skull fracture with intracranial hemorrhage*
  - *Skull fracture without intracranial hemorrhage*
    - Presence/absence intracranial hemorrhage
      - May change the course of the disease of which skull fracture is a part
      - Does not change the skull fracture itself
- Clinical terminology systems have to provide epistemological features

Epistemology in biomedical ontology

- Biomedical ontologies do NOT have to provide epistemological features
  - But clinical terminology services based on bioledical ontologies would have to
- If epistemological features are present in biomedical ontologies, they should be clearly recorded as such
- Epistemology must be recognized as something important by the biomedical ontology community

Unresolved issues

- What constitutes a biomedical class?
  - Classes defined as the complement of other classes
    Non-insulin dependent diabetes mellitus
  - *Not otherwise specified* is to be banned
  - What about *Not elsewhere classified*?
- What is biomedical reality?
- What is normality?



Medical Ontology Research

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